Tanya M. Harding, Ph.D.
Our Ref. No. 4239-67289
Express Mail Label No. EV339203299US
FOR: ATP-BINDING TRANSPORTER (ABC7) AND METHODS
FOR DETECTION OF ANEMIA AND ATAXIA
Inventor(s): Michael C. Dean, Amy Ann Hutchinson and
Rando Lembit Allikmets
Date of Deposit: January 21, 2004

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| ABC7 | QIP | E S | L | K S | I 7 | r v | I Q | R | L | G I | K (| 3 N | S | G | Q | F: | L | D A | . A | K | ΑI | Ď | VV | N E | L | I | Е | K I | RT | C | W | H | G H | ΙA | G | G | G 1 | L Ի | ł | |
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| Atmlp | TSK | ΑP | T : | LS | ΕI | ٠. | | | K | I | L I | K D | L | F | R | Y | I١ | W P | K | G I | 1 1 | 1 K | V | R 1 | R | V | L | I | A L | G | L | L | I S | S A | K | I | Ll | 7 V | 7 | |
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| Atm1p | QVP | FF | F | ΚQ | T 1 | [[| S | M | N | | | | | | I | A ! | W I | ם ס | P | Т ' | V Z | \ L | P | A A | I | G | L, | r | ΙL | C | Y | G | V A | A R | F | G | s v | VI | _ | |
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| Atm1p | GTK | GΙ | S | QΨ | ГL | r <i>P</i> | M | v | F | н : | Ι: | ΙP | I | s | F | E | I | s v | v | C (| G I | L | T ? | YÇ | F | G | A | S I | FA | A | I | T | F S | Т | M | L | L? | Y S | 3 | |
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| | 350 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | М | | | | | |
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| | | | | | | | | | _ | | M6 | | | _ | _ | | | | _ | | | | | • | | | | | | | | | | | | | | | | |
| | 450 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | ` | | | | | | | | |
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| Atm1p | KN. | . A | E I | R P | L | 1 I | , P | E | N | V I | P : | Y D | Ι | T | F | E | N ' | VΊ | F | G ' | Y F | ΗP | DI | R F | ί | L | K | N Z | A S | F | Т | I | P A | A G | W | ĸ | T | A I | [| |
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| | 499 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| ABC7 | VGG | SG | S | GΚ | S | 1 | : V | R | L | L | FI | R F | Y | E | P | Q: | K (| 3 S | I | Υ : | LA | A G | Q I | I | Q I | D | v | S | LΕ | s | L | R | R A | V A | G | v | V I | Pζ | 2 | |
| Atm1p | VGS | ·S G | S | GΚ | S | . 1 | L | K | L | V I | F | R F | Y | D | P | E | S | G R | Ι | L: | 1 | 1 G | RI | נכ | K | E | Y: | D : | I D | Α | L | R | ĸν | <i>7</i> I | G | v | V I | Pζ | 2 | |
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| | 549 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
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| | 599 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| ABC7 | ERG | ЬK | L | SG | GI | E F | Ç | R | V | Α: | Ι / | A R | Α | Ι | L | K | D I | PΡ | V | I | L > | Y D | E A | 47 | S | S | L | D S | SI | T | E | E ' | T I | L | G | Α | M I | ΚI |) | |
| Atmlp | ERG | LM | I | SG | GI | E F | Q | R | L | A : | I | A R | v | L | L | K | N Z | A R | Ι | M I | FE | P D | E A | A 7 | S | Α | L | D ? | ГН | T | E | Q. | ΑI | L | R | T | I | RI |) | |
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| | 649 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
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| Atm1p | NFT | SG | S | RT | S | / 3 | Ί | Α | Н | R I | LI | R T | Ι | A | D | A I | D I | ΚI | Ι | V: | LI | ИС | G I | R 1 | R | E | E | G 1 | K H | L | E | L | LP | M | P | G | S | լ չ | Č | |
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FIGURE 1A

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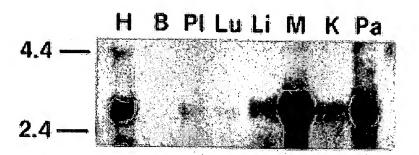
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|------|------------|------------|------------|------------|--------------|
| 1 | ATGGCGCTGC | TCGCGATGCA | TTCTTGGCGC | TGGGCGGCCG | CGGCGGCTGC |
| 51 | TTTCGAAAAG | CGCCGGCACT | CCGCGATTCT | GATCCGGCCT | TTAGTCTCTG |
| 101 | TTAGCGGCTC | AGGTCCGCAG | TGGAGGCCAC | ATCAACTCGG | CGCCTTGGGA |
| 151 | | CCTACCAGAT | | TTAAAAAGTA | |
| | | | | | |
| 201 | | AAAGGCAATT | | | |
| 251 | TCCAGGTATG | GCCACTGATA | GAAAAGAGGA | CATGTTGGCA | TGGTCATGCA |
| 301 | GGAGGAGGAC | TCCACACAGA | CCCAAAAGAA | GGGTTAAAAG | ATGTTGATAC |
| 351 | TCGGAAAATC | ATAAAAGCAA | TGCTTTCTTA | TGTGTGGCCC | AAAGACAGGC |
| 401 | CAGATCTACG | AGCTAGAGTT | GCCATTTCGC | TGGGATTTTT | GGGTGGTGCA |
| 451 | | ATATTGTGGT | | TTTAAATATG | CTGTAGACAG |
| 501 | | ATGTCGGGAA | | | |
| | | | | | |
| 551 | | CATGGCAACA | | | TGTATCAAGA |
| 601 | GCTGGAGCTG | CTTTTTTTAA | CGAAGTTCGA | AATGCAGTAT | TTGGCAAGGT |
| 651 | AGCCCAGAAT | TCAATCCGAA | GAATAGCCAA | AAATGTCTTT | CTCCATCTTC |
| 701 | ACAACCTGGA | TCTGGGTTTT | CACCTGAGCA | GACAGACGGG | AGCTTTATCT |
| 751 | | ACAGAGGAAC | | | TGAGTGCTTT |
| 801 | | CTTCTTCCCA | | | GTCAGTGGTG |
| 851 | | | | | |
| | | CAAATGCGGT | | | CCTTGGAACA |
| 901 | | ACACAGCATT | | | |
| 951 | ATTTAGAATA | GAAATGAACA | AAGCAGATAA | TGATGCAGGT | AATGCTGCTA |
| 1001 | TAGACTCACT | GCTGAATTAT | GAAACTGTGA | AGTATTTTAA | TAATGAAAGA |
| 1051 | TATGAAGCAC | AGAGATATGA | TGGATTTTTG | AAGACGTATG | AGACTGCTTC |
| 1101 | | ACCTCTACTC | | | |
| 1151 | | CGGTTTAACA | | | |
| 1201 | | | | | |
| | GTGGCAGGTA | | | GTAATGGTGA | |
| 1251 | TTTTCAGCTT | TCATTACCCC | _ | GGGAACTGTA | |
| 1301 | | ACTCATAGAT | | | |
| 1351 | GACACCCAAA | TTAAAGACAA | AGTGATGGCA | TCTCCCCTTC | AGATCACACC |
| 1401 | ACAGACAGCT | ACCGTGGCCT | TTGATAATGT | GCATTTTGAA | TACATTGAGG |
| 1451 | GCCAGAAAGT | CCTTAGTGGA | ATATCCTTTG | AAGTCCCTGC | AGGAAAGAAA |
| 1501 | | TAGGAGGTAG | | | |
| | ATTATTTCGC | | | | |
| | | | | | - |
| | AAAATATACA | | | | _ |
| | GTACCTCAGG | | | | |
| | ATATGGAAAC | | | | |
| 1751 | TAGCTGGACT | TCATGATGCA | ATTCTTCGAA | TGCCACATGG | ATATGACACC |
| 1801 | CAAGTAGGGG | AACGAGGACT | CAAGCTTTCA | GGAGGAGAAA | AGCAAAGAGT |
| 1851 | AGCAATTGCA | AGAGCCATTT | TGAAGGACCC | CCCAGTCATA | CTCTATGATG |
| | AAGCTACTTC | | | | |
| | ATGAAGGATG | | | | |
| | | | | | |
| | GTCAACAGTG | | | | |
| | TAGCCGAACG | | | | |
| 2101 | TATTCAGAAA | TGTGGCATAC | ACAGAGCAGC | CGTGTGCAGA | ACCATGATAA |
| 2151 | CCCCAAATGG | GAAGCAAAGA | AAGAAAATAT | ATCCAAAGAG | GAGGAAAGAA |
| 2201 | AGAAACTACA | AGAAGAAATT | GTCAATAGTG | TGAAAGGCTG | TGGAAACTGT |
| | TCGTGCTAAG | | | | |
| | TATTTGCACT | | | | |
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| 1 | ATGGCGCTGC | ŤCGCGATGCA | ттсттссссс | TGGGCGGCCG | СССССССТСС |
|------|------------|--------------------------|------------|------------|--|
| 51 | | CGCCGGCACT | | | |
| 101 | | AGGTCCGCAG | | _ | |
| 151 | | CCTACCAGAT | | TTAAAAAGTA | |
| 201 | | AAAGGCAATT | | | |
| 251 | | GCCACTGATA | | | |
| 301 | | TCCACACAGA | | | |
| 351 | | ATAAAAGCAA | | | |
| 401 | | AGCTAGAGTT | | | |
| 451 | | ATATTGTGGT | | TTTAAATATG | CTGTAGACAG |
| 501 | | ATGTCGGGAA | | · = | - - - |
| 551 | | CATGGCAACA | | | TGTATCAAGA |
| 601 | | CTTTTTTTAA | | | |
| 651 | | TCAATCCGAA | | | |
| 701 | | TCTGGGTTTT | | | |
| 751 | | ACAGAGGAAC | | | |
| 801 | | CTTCTTCCCA | | | GTCAGTGGTG |
| 851 | | CAAATGCGGT | | | CCTTGGAACA |
| 901 | | ACACAGCATT | | | |
| 951 | | GAAATGAACA | | | _ |
| | | GCTGAATTAT | | | |
| 1051 | | AGAGATATGA | | | |
| | | ACCTCTACTC | | | |
| | | CGGTTTAACA | | | |
| | | CCCTTACTGT | | | |
| | | TCATTACCCC | | | |
| | | ACTCATAGAT | | • | |
| | | TTAAAGACAA | | | |
| | | | | | |
| | | ACCGTGGCCT CCTTAGTGGA | | | |
| 1501 | | | | | |
| | | TAGGAGGTAG | | | |
| | | TTCTATGAGC | | | |
| | | AGATGTGAGC ATGCTGTCCT | | | |
| | | ATCAGTGCTT | | | |
| | | TCATGATGCA | | | |
| | | AACGAGGACT | | | |
| | | AGAGCCATTT | | | |
| | | | | | |
| | | ATCGTTAGAT | | | |
| | | TGGTCAAACA | | | |
| | | GTTGATGCAG | | | |
| | | TGGTACCCAC | | | |
| | | TGTGGCATAC | | | |
| | | GAAGCAAAGA | | | |
| | | AGAAGAAATT | | | |
| | | TCACATAAGA | | | |
| 2301 | IMITIGCACT | GAAGCAGAAT | IGITITALIA | AAAAAATCAT | ACAII |

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FIGURE 2



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FIGURE 3

